SPRING 2024

INSIDE

FSIG Connection

News from the Fabry Support & Information Group

Kidney recipient returns to job

Update on NC music teacher, who also got hips replaced

COURTESY OF BRYAN LONG

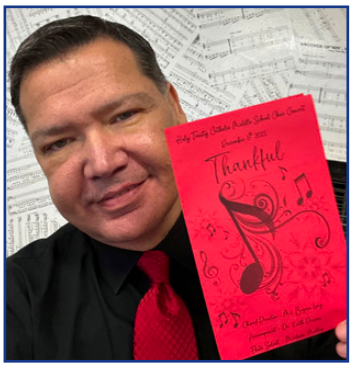
Long directs a middle school Christmas concert in Charlotte, NC.

By Melissa James
FSIG Contributor

Back to work—a grind for some, but for Fabry patient Bryan Long, it's music to his ears.

FSIG Connection first started following Long's story back in 2018, when we reprinted an article about him from his local newspaper, *The Charlotte Observer*. At that time, the North Carolina music teacher had been publicly seeking a kidney donor.

After a friend posted Long's plea on Facebook, a woman who lived nearby got tested and was a match.



Natalie Korda, a fellow educator who was teaching Spanish in a different school division, gave him a kidney and a new chance at life.

The two became fast friends, and a grateful Long was now free from dialysis. But his journey wasn't over. The transplant surgery brought weight gain, which prompted Long to jump on the exercise bike two to three times per day, riding up to 5 miles at a stretch.

Between the weight and the bodily wear, Long's hips developed avascular necrosis—death of bone tissue due to a lack of blood supply, which Long learned is somewhat common among Fabry patients. For over a year, Long was unable to walk without assistance. He underwent a double hip replacement in late 2022. The following August, he reentered Holy Trinity Catholic Middle School as its music director and choir teacher.

"I did not know for sure if I was physically capable of returning to work, but I felt I

See LONG, Page 2

Man's transplant a success, but crises continue

By Allen Neally
Fabry Patient

This is my story of living with Fabry disease. In my childhood, anytime I got sick, whether it was an ear infection, or cold, etc., I had such burning and amazing neuropathy that I would cry and beg for it to stop.

I can remember my mother coming in and sitting with me all hours of the night. Even though she was mostly asymptomatic, she had had these episodes in the past and knew what I was going through. It must have been frustrating for both my mother and father knowing that they didn't know why this happened, but only could console me. I struggled through those years with a lot of pain, discomfort, and sickness.

When I was 13, I spent a week at Thayer Hospital in Waterville, Maine. Neurologists performed tests, spinal taps and lots of blood work. Doctor's diagnosis: idiopathic (disease with no identifiable cause).

As I moved through my early teens, I also developed lymphedema—constant swelling of the hands and feet. It was very embarrassing to me.

See CRISES, Page 3

WELCOME

Dear Friends,

This issue of FSIG Connection highlights several Fabry kidney-transplant recipients and their stories of endurance despite many physical challenges. We also continue our ongoing look at legislation for the removal of co-pay accumulators. And as always, we spotlight the latest progress in Fabry medical research and ongoing clinical trials.

We also are happy to share a recap of the 2024 FSIG Expert Fabry meeting held in San Diego, CA, and next issue we will share an update of the latest T4T fundraiser and our annual Fabry Fun Run Walk.

Jack

Jack Johnson,
Executive Director

Kidney disease in Fabry patients more common since 2000

Over the past two decades, kidney disease and urinary tract problems have become more common among people with Fabry disease, especially among female or Black patients.

In this study, scientists in China analyzed data from 10,637 people with Fabry disease in the U.S. who were treated between 2000 and 2020. The scientists compared rates of kidney disease and related problems among patients in the first decade (2000 to 2010) to the second (2010 to 2020).

"Of particular note, the prevalence of advanced chronic kidney disease stages 4 and 5 [severe or end-stage disease] nearly doubled over the study period," the researchers wrote.

Read full story: bit.ly/Fabry-Kidney

LONG: continued from page 1

had to give it a try," he said. "I felt like I still had so much to contribute and there is an intended purpose that I was given a second chance. I believe the timing was perfect!"

His co-workers were thrilled to have him back.

"It was very much like somebody was missing from the family. So, we were very glad, very joyful for his return," Principal Kevin Parks told Spectrum News. Prior to his administrative role, Parks taught alongside Long at the middle school.

Long said he's happy to be back at the school that never forgot him, and to share his passion with a new crop of students. What's it been like teaching post-pandemic for the first time?

"The experience of live music, performing and concert etiquette are all foreign to most of these students because of all the restrictions and distancing during Covid. They all seem to be willing to learn and have done very well in working toward the goals we have set."

He added that his students are very aware of his health condition and are careful to avoid spreading germs.

Long called his new hips "wonderful" and said he makes sure to move frequently, alternating between time at the piano and standing so his hips can stretch and strengthen daily. As is common for a transplant patient, there are good days and challenging days for Long.

COURTESY OF BRYAN LONG

Long with Natalie Korda, his kidney donor, who came out to cheer and support his first school concert back at work.

COURTESY OF BRYAN LONG

Social media post from a former student.

"I'm so thankful to be alive and to have the opportunities to teach and share my passion for music, that it seems wrong to ever complain," he said. "My overall health is good, my spirit is encouraged by the people who I teach as well as others, and my least favorite day at work is far greater than any day on dialysis. I feel grateful."

Long also wanted to share these words of comfort to his fellow Fabbers:

"Only people who are affected by this disease can truly understand the struggles we face. Reach out to this newfound virtual community of Fabry patients to voice concerns or find information that can assist in treatment, financial resources and overall mental health as well as physical.

Be kind to yourself. Try not to take it personal when people don't understand the pain, the frustration and the emotional impact of this disease and dismiss you because you 'don't look sick.' You can make a difference! You can encourage someone who may need encouragement. You can ALWAYS choose kindness."

KIDNEY BRIEFS

Mutation tied to Fabry in woman with kidney disease

(Dec. 29, 2023) A mutation in the GLA gene associated with Fabry disease was found by Italian researchers in a woman with end-stage kidney disease, according to a case report study.

The 52-year-old woman was the sole family member with the disease-causing variant, meaning it wasn't inherited from her parents. Her diagnosis occurred later in life.

"The detection of a de novo mutation in just one family member is a rare occurrence that highlights the importance of genetic counseling, early diagnosis, and, finally, extended genetic screening for Fabry disease in patients affected by [kidney] disease of unknown etiology," the researchers wrote.

Read full story: bit.ly/Fabry-Mutation

Woman has 2 disorders affecting heart, kidneys

(Feb. 23, 2024) A woman with Fabry disease was found to have two co-occurring disorders — a heart condition called dilated cardiomyopathy (DCM) and an autoimmune kidney disorder called immunoglobulin A nephropathy (IgAN) — a rare combination.

Scientists in China described the case of a 60-year-old woman with all three conditions. The woman sought medical attention for swelling in her face and legs, and chest tightness when walking. Genetic testing revealed the woman had a Fabry-causing mutation in the GLA gene and DCM-associated mutations in both the TTN and BAG3 genes. Her 35-year-old son was found to carry the same Fabry-causing GLA mutation.

Read full story: bit.ly/Co-Disorders

Screening for Fabry vital in chronic kidney disease

(Feb. 2, 2024) Among people receiving medical care for chronic kidney diseases, a small fraction have Fabry disease, a new study highlights.

To ensure these patients get optimal treatment, it's critical to have screening programs in place, according to researchers.

"Although the overall prevalence of [Fabry disease] is low in patients with kidney involvement, screening, especially in patients who have not yet undergone kidney replacement therapy, is important, in order to provide timely and effective treatment interventions," the scientists wrote in the study, "Prevalence of Fabry disease in patients with chronic kidney disease: A systematic review and meta-analysis."

Read full story: bit.ly/Fabry-CKD

CRISES: continued from page 1

When I was 17, I was sent to Maine Medical Center in Portland and put through a battery of tests that lasted over a week. That visit happened after I injured my wrist. My parents had brought me to a friend of the family, an orthopedic surgeon, who took X-rays. Nothing was broken, but he noticed I had lots of space between the joints in the fingers allowing for growth, yet I wasn't growing. I also was not maturing physically. It was then they determined I had an inactive pituitary gland.

They gave me testosterone injections for about a year and a half, which helped me put on some weight and some height. I was thankful. What the injections did not do was stop any of my other problems—neuropathy, lymphedema and a continued state of not feeling well.

As I moved into my adult years, the neuropathy seemed to dissipate except for when I got sick. The lymphedema also become more moderate. I found that 100 mg carbamazepine chewables really helped during temperature fluctuations in spring and fall temperatures.

In my early 30s, I went down to the Lahey Clinic in Massachusetts and started all over again with batteries of questions and tests. At the end of a few days, the only thing they could find was that my salt content looked a bit high (genius). Well, I was staying next door in a hotel

eating things like prime rib and French onion soup... I don't suppose that had anything to do with it!

They suggested a low-salt diet, and I went home with no change.

A few years later, I had an appointment with my primary care physician. He took my vitals, and my blood pressure was around 220 over 160. He referred me to a local nephrologist, who also was confused by the high blood pressure. He took a biopsy of my kidney and sent it t

o Mayo Clinic. Two weeks later, I was diagnosed with Fabry disease.

This gave me so much clarity and a sense of relief—

COURTESY OF ALLEN NEALLY

Allen Neally finally got a Fabry diagnosis after years of unexplained symptoms.

no longer being accused of saying I was sick without explanation. But despite the good news of a diagnosis, my kidneys were not faring so well. Four years later, I was in renal failure and needed a kidney transplant.

Fortunately, my sister was a perfect 6 antigen match, and the transplant surgery was a success.

But my troubles aren't over. This disease is systemic: It's affecting my heart, I've lost my hearing, I battle constant abdominal cramping and I have severe Fabry crises anytime I get an infection. The crises have actually become my early indicator that an infection is starting, before any bloodwork is done to confirm it.

When I'm in the midst of a Fabry crisis, the only thing that helps is strong pain medication—which the medical industry now frowns on. So I have difficulty getting relief. These crises are so debilitating that I don't even want to move. So I don't.

I only get the fluids that I need for medications and get to the bathroom. I don't eat, and I become severely dehydrated and malnutritioned, which brings their own set of problems.

As I grow older, the pain grows stronger and lasts longer. Trying to get physicians to understand my need for pain medication is no easy task, and usually it's only going to happen in a hospital.

Fabry kids can benefit from Make-A-Wish

By Susanna VanVickle
FabryDiseaseNews.com

(Oct. 17, 2023) My twin sons, Anthony and Michael, were diagnosed with Fabry disease at age 17.

The Make-A-Wish Foundation is an extraordinary organization that brings joy to suffering children by making their wishes come true. I'd originally thought it only granted wishes for terminally ill children, but I've since learned these once-in-a-lifetime opportunities go to thousands of kids with diagnoses that require lifelong medical attention.

Both boys quickly qualified with Make-A-Wish, which was a blessing that boosted the morale of our whole family.

Being appreciative and pragmatic, Anthony wished for money for college. That was a good choice during a time when COVID-19 had slowed down many types of wish-granting, and especially so since he was about to attend Benedictine College in Kansas.

The North Texas Make-A-Wish volunteers were amazing. When Anthony's wish was granted (and a check was mailed to his college), a lovely wish-granter came to our home with dinner and sweet treats for the whole family!

Throughout the process, Anthony received so much more than the college money. He got cards and gifts from Make-A-Wish, a chance to have Chick-fil-A deliver a meal to the family, and other gift cards and bonus goodies. Most important, he was able to delight in all these gifts because of his disease, not in spite of it.

Michael, unlike his more practical twin brother, is a dreamer. He'd long had a thing for old pickup trucks, and he'd begun to envision having a classic. So he bought a 1979 Chevrolet C20, and his wish was to have the interior restored.

A group of five, all in shorts, stand in front of a blue and white, extra-large truck, all in front of a couple of old gas pumps. He got the old truck inexpensively because the interior was pathetic and the engine needed work. He spent countless hours with his brothers and friends working on the truck, with more trips to the mechanic shop than I care to recount.

Make-A-Wish grants wishes to kids 18 or younger, but Michael was 20 when his wish was completed. During the three years he waited, he enjoyed T-shirts, swag, and other small gifts from the foundation. He also loved having his precious truck as a conversation piece and spent an invaluable amount of time bonding with his brothers and friends as they worked on it.

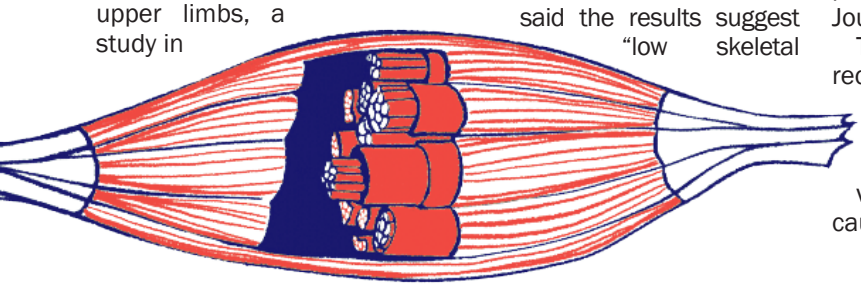
The day of his wish-granting didn't come quickly, but it was worth the wait.

Read full story: bit.ly/FabryWish

Low muscle mass may be early sign of Fabry in children

By Andrea Lobo, PhD
FabryDiseaseNews.com

(Nov 10, 2023) Most children with Fabry disease, particularly those with the severe, classic form, have lower than normal skeletal muscle mass that affects the lower limbs more than the upper limbs, a study in China shows.



Skeletal muscles are responsible for voluntary movements and play a vital role in everyday activities.

"This is the first study to examine body composition and muscle mass in early FD [Fabry disease] patients," wrote the researchers, who said the results suggest "low skeletal muscle mass may be one of the early manifestations of FD."

"Physicians need to pay close attention to this aspect in children with FD," the researchers wrote in "Low skeletal muscle mass as an early sign in children with fabry disease," which was published in the Orphanet Journal of Rare Diseases.

The Fabry-associated reduction of muscle mass might be due to the direct destruction of muscle fibers and/or blood vessels in the muscles, caused by the abnormal deposition of fatty molecules, the researchers said, noting "reduction in muscle mass may also be involved in the development of osteoporosis in FD patients, since there is solid evidence that low muscle mass is associated with a reduction in bone parameters during growth and increases the risk of osteoporosis in adulthood." In this study, four boys with classic Fabry had lower than normal bone mass.

More research with larger sample sizes is needed, said the researchers.

Read full story: bit.ly/FabryMM

UK team seeking to boost early diagnosis in minorities

By Lindsey Shapiro, PhD
FabryDiseaseNews.com

(March 22, 2024) Researchers in England are working toward identifying and addressing barriers to reaching a Fabry disease diagnosis among underrepresented communities, particularly within ethnic minority and low socioeconomic status groups.

The project is spearheaded by Richard Steeds, MD, a professor at the University of Birmingham and consultant cardiologist at University Hospitals Birmingham. Amicus Therapeutics, developer of approved Fabry therapy Galafold (migalastat), will provide some funding for the project.

"If we can address these barriers, then we can support more people to benefit from beginning treatment for this rare condition sooner," Steeds said in a university press release.

As with most progressive diseases, early treatment of Fabry disease is key for slowing disease progression and offering patients better outcomes. Approved therapies, including enzyme replacement therapy and chaperone therapy, can help to slow the progression of organ damage and ease Fabry symptoms.

To start treatment early requires a prompt diagnosis. Since Fabry disease is inherited, family screening is a critical tool for catching cases early on.

However, because Fabry disease is quite rare, it is easily missed. Patients commonly experience diagnostic delays or are misdiagnosed with more recognizable conditions. Indeed, research suggests the disease remains significantly underdiagnosed.

Such issues may be amplified among minority ethnic groups, despite the fact that Fabry disease is found roughly equally in all racial and ethnic groups. Language barriers, low health literacy, and cultural and other factors within minority communities may contribute to stigma that makes individuals less likely to seek care or family screening.

In turn, patients may be diagnosed later and have worse health outcomes, contributing further to health inequities in these groups.

"Previous work has shown that Black, South Asian, and other minority ethnic groups, as well as patients from lower socioeconomic groups, are underrepresented in our clinic in Birmingham, despite it being a very diverse city," Steeds noted.

"This knowledge helped us to identify a need for further research to understand and overcome specific barriers to family screening that are more prevalent within minority ethnic groups."

To address these disparities, the team is working on building advisory groups consisting of community leaders, healthcare professionals, and patients from Black, South Asian, and other minority groups.

The goal is for these groups to help the researchers better understand the barriers patients face and what strategies could be used to overcome them. Once they've identified possible approaches to do so via the advisory groups, the scientists will test them out in their home city of Birmingham, but they'll first need to secure additional funding.

Read full story: bit.ly/3TEToYU

Online games help parents detect LSDs

By Tiziana Moriconi
LatestBreakingNews.com

(Jan 2, 2024) Can you find the hidden object, find the differences or the shortest way out of the maze? It is not the puzzle week, but the puzzles of Bravo chi trova, the original format of the third edition of the 'Raro chi trova' campaign which, with games of logic and intuition, aims to help parents get on the trail of lysosomal storage diseases in the belief that even if the disease is rare you can find it. And noticing the signs of these diseases early can really make a difference for young patients and their families.

Precisely because early diagnosis is fundamental, parents have the arduous task of recognizing the signs, which is not an easy task. Yet, with a little attention and training it is possible to find a clue and get on the trail of these diseases.

Play and discovery are the common thread of the three editions of "Raro Chi Trova," an awareness campaign on Fabry and Gaucher diseases and Hunter syndrome.

"Informing parents how this awareness campaign is proposed," observed Stefania Tobaldini, president of the Italian Anderson-Fabry Association, "is essential to increase knowledge and is the first step towards awareness to discover Fabry disease early. Knowing if your child has a rare pathology helps the family to face and not suffer its consequences, to bear the burden from an emotional and management point of view, avoiding wandering from one doctor to another."

Play the games in English or Italian:
www.rarochitrova.it

TRIAL BRIEFS

More China trials needed to fill Fabry "treatment gap"

(Sept. 29, 2023) The current treatment landscape for Fabry disease in China is completely dependent on enzyme replacement therapies (ERTs) developed by western companies like Sanofi (Fabrazyme) and Takeda (Replagal).

Due to the limited affordability of these therapies, there exists a substantial unmet clinical need that could be addressed by newer therapies. However, only a few western innovator companies conducted clinical trials for new Fabry disease pipeline therapies in China, despite the country having the highest number of diagnosed prevalent cases of Fabry disease among the 16 major markets (16MM*), says GlobalData, a leading data and analytics company.

Read full story: bit.ly/Treatment-Gap

Elfabrio slows kidney decline in Phase 3 trial

(Jan. 19, 2024) A year of treatment with Elfabrio (pegunigalsidase alfa) is well tolerated and slows the progression of kidney disease in men and women with Fabry, according to final published data from the BRIDGE Phase 3 clinical trial.

The ERT from Protalix BioTherapeutics and Chiesi Global Rare Diseases was approved in the U.S. last year for adults with Fabry disease, backed in part by data from this trial.

Formerly known as PRX-102, Elfabrio is designed to deliver a functional version of the Gal A enzyme to Fabry disease patients. It was developed using Protalix's plant-based platform and is expected to last longer in the bloodstream than other ERTs.

The open-label BRIDGE clinical trial (NCT03018730) set out to evaluate the safety and efficacy of Elfabrio, delivered via into-the-vein infusions (1 mg/kg) once every two weeks, over a year in 22 adults with Fabry disease who had been previously treated with Replagal (agalsidase alfa; 0.2 mg/kg) for at least 2 years.

Read full story: bit.ly/Elfabrio-Slows

Phase 1 trial of AL01211 oral shows effective, safe results

Phase 2 trial results likely later this year

By Lindsey Shapiro
FabryDiseaseNews.com

(March 4, 2024) The investigational therapy AL01211 safely lowered levels of globotriaosylceramide (Gb3)—the molecule that toxically accumulates in Fabry disease—among healthy adults taking part in a Phase 1 trial, according to published results.

The oral treatment, being developed by AceLink Therapeutics, also is being tested in a Phase 2 clinical trial (NCT06114329) in about 16 men with classic Fabry disease, ages 18-60, who have never used a disease-related treatment. Recruitment may be continuing at six sites across China.

Top-line Phase 2 trial results are anticipated in the second half of this year.

"These data reinforce our commitment to provide more convenient and more effective therapeutic options to patients suffering from glycosphingolipid storage diseases such as Fabry disease," Michael Babcock, PhD, vice president of research and early development at AceLink, said.

"The insights gained from this study have set us up nicely to test the therapeutic benefits of AL01211 in patients," Babcock said.

The AceLink-supported study, "Phase 1 Healthy Volunteer Study of AL01211, an Oral, Non-brain Penetrant Glucosylceramide Synthase Inhibitor, to Treat Fabry Disease and Type 1 Gaucher Disease," was published in *Clinical Pharmacology In Drug Development*.

Investigational SRTs for Fabry, like venglustat and lucerastat, aim to lower Gb3 production by inhibiting glucosylceramide synthase (GCS), an enzyme involved in the synthesis of a group of fatty molecules called glycosphingolipids. Gb3 belongs to this fatty molecule group.

AL01211 is a novel SRT that's designed to avoid entering a patient's central nervous system (the brain and spinal cord) as it works to block GCS. In this way, it aims to maximize its effects on targeted organs and minimize any side effects on the brain.

AceLink is developing AL01211 as an oral treatment for Fabry disease and Gaucher disease type 1, which is characterized by the accumulation of a different type of glycosphingolipid, one called glucosylceramide (GL1).

Read full story: bit.ly/Al01211-results

FDA: Get another study for gene therapy approval

By Marisa Wexler, MS
FabryDiseaseNews.com

(Feb. 16, 2024) The U.S. Food and Drug Administration has advised Sangamo Therapeutics that positive results from a single well-controlled clinical trial, in addition to existing evidence, may be sufficient to form the basis for approving its Fabry disease gene therapy candidate.

For Sangamo, that means that one more study may be enough for a positive regulatory decision in the U.S. on its ST-920 (isaralgagene civaparvovec) gene therapy—which "would significantly reduce anticipated complexity, cost and time to potential approval," the company said in a press release.

In a meeting with the developer, the FDA agreed that the proposed trial would include up to 25 male and female participants with Fabry disease. The agency also agreed that the trial would not require a control arm or a head-to-head comparison with a standard enzyme replacement therapy (ERT), which is expected to allow the study to be done faster and more cheaply.

"We are thankful for the FDA's support and alignment on a regulatory pathway that could potentially deliver a new treatment option for Fabry disease patients on an expedited, cost-effective timeline," said Nathalie Dubois Stringfellow, PhD, chief development officer.

On the other side of the Atlantic, the European Medicines Agency (EMA) has granted ST-920 priority medicines (PRIME) eligibility, a designation given to therapies with the potential to fill unmet medical needs that aims to get much-needed treatments to market faster.

The FDA also has given the Fabry gene therapy candidate fast track designation, which hastens the review of potentially important therapies, as well as regenerative medicine advanced therapy designation under the 21st Century Cures Act.

Read full story: bit.ly/1MoreStudy

6 things RD patients want clinical trial sponsors to know

In a Jan. 9 guest column on ClinicalLeader.com, rare disease patient, caregiver and advocate Sabina Kineen shared her advice to clinical trial sponsors. Kineen, who like her father was diagnosed with Fabry, based these insights on her experience partnering with a sponsor—where she got to see firsthand what goes into a trial design and execution.

She went from being a trial participant to "speaking at late-cycle meetings to share the patient voice." Her key takeaways for sponsors :

1. Understand that every single trial participant is different. Get to know what each of them needs in their life to make their participation as seamless as possible. Ask them what their burden is and what you can do to lift it.

2. Understand the disease and its limitations on your patient participants. For example, as a Phase 2 participant, I endured a battery of tests—yet as a Fabry patient, I struggle with fatigue, which sometimes leads to a pain crisis. This made it difficult to adhere to the back-to-back timing of the tests.

3. Save patient travel time and study cost by incorporating decentralized elements. Consider using remote data collection, mobile research units, and/or patients' local lab facilities for things like patient histories, blood draws, etc. Not everyone can take multiple days off work.

4. Communicate trial information with community groups. With the help of patient organizations, sponsors have made great strides in both patient experience and, even before that, in educating the public that there are even trials going on. This has been facilitated by the realities of modern communication.

5. Improve trial diversity by working with community leaders and partnering with former trial participants. Those from underserved communities who might not otherwise discuss medical issues will feel more comfortable seeing someone who looks like them, is in their season of life and has been through it.

6. Aim to alleviate both patient and caregiver burden. For patients who have someone supporting them throughout the course of your clinical trial, the time, energy, and cost affects both lives and may need to be uniquely addressed for each person—especially if they don't live together.

Read full story: bit.ly/4cTl5pJ

FSIG

FSIG is a support group dedicated to dispensing information and encouraging mutual self-help as a means of emotional support.

FSIG was formed in 1996 by two Fabry patients and supportive family members with the hope that their particular understanding of this disease, combined with experience gathering information and working with doctors could benefit others.

FSIG is a nonprofit, tax-exempt organization and relies on charitable contributions to provide services to those with Fabry disease, their families and supportive others. Donations may be sent to the address below.

Please feel free to make copies of the FSIG Newsletter to share with your family, friends and others. We encourage anyone interested in FSIG or the newsletter to contact us so we can make sure you receive the next issue.

FSIG
108 NE 2nd St.
P.O. Box 510
Concordia, MO 64020-0510

Phone: (USA) 660-463-1355
Toll-Free: (USA) 866-30-FABRY
Fax: (USA) 660-463-1356
Web: www.fabry.org
Email: info@fabry.org

FOLLOW US!
LIKE US!

Guidelines for using Galafold focus on patient empowerment

By Marisa Wexler, MS
FabryDiseaseNews.com

(Oct. 6, 2023) A team of expert clinicians and patient advocates has created a new set of recommendations to guide the use of Galafold (migalastat) in people with Fabry disease, highlighting the importance of centering patients' preferences in treatment decisions.

Here a panel of 16 experts—14 physicians and two patient advocates—created a set of recommendations for Galafold in Fabry disease. To create the guidelines, the panel used a Delphi process, where they voted on a series of statements in two rounds, with refinement of the statements after the first round.

The guidelines suggest that, when Fabry disease is diagnosed, clinicians should have comprehensive discussions with patients about what their treatment options are, including eligibility for Galafold based on mutation status.

If the decision is made to start on Galafold, patients should undergo a comprehensive set of evaluations to assess for Fabry-related signs and symptoms, including measures of pain, digestive issues, and markers of heart, kidney, and brain health. Mental health and life quality assessments also should be measured when treatment is started.

These measures should then be repeated again about three months after starting on Galafold, and then again at regular intervals for the duration of treatment.

By conducting regular monitoring, clinicians and patients can increase the odds that, if a new or worsening issue arises, it can be detected early so appropriate care can be started in a timely manner.

The panel recommended clinicians should regularly check alpha-gal A enzyme activity and measure lyso-Gb3 levels, but said these biomarkers should not be the only thing guiding treatment decisions, emphasizing also the importance of assessing organ function and symptom severity when deciding whether to continue, stop, or switch treatments.

"Treatment decisions should take all measured parameters into account, including all Fabry-related symptoms, signs of organ involvement, and pharmacodynamic biomarkers," the researchers wrote.

The recommendations also underscore the importance of centering the patient's wishes and preferences in discussions about treatment decisions, and having honest discussions about what is and is not known.

"Where research is lacking, discussion of the available evidence between healthcare professionals (HCPs) and patients can address doubts and empower the patient in the process," the team wrote.

The guidelines note that if lyso-Gb3 levels increase unexpectedly, clinicians should talk to patients to ensure they're taking Galafold as prescribed.

"Further research is required to investigate current and potential biomarkers (including [alpha]-Gal A and lyso-Gb3 but not limited to substrate biomarkers) and determine any other potential prognostic tools," the scientists said. The researchers noted that, while these recommendations focused on Galafold only.

Read full story:
bit.ly/Galafold-Guidelines

Study: Fabrazyme infusion time safe to reduce

With care, higher infusion rates had no impact on patient safety

By Andrea Lobo, PhD
FabryDiseaseNews.com

(Sept. 29, 2023) The duration and rate of Fabrazyme (agalsidase beta) infusion can be safely changed in people with Fabry disease, independently of their body weight, if appropriate care is taken, according to a new study from Japan.

Specifically, an infusion time shorter than 90 minutes or an infusion rate higher than 15 mg/hour—both standard procedure—had no significant impact on treatment safety.

"These findings suggest infusion times in patients who are tolerating treatment can, with careful monitoring, be gradually decreased," the researchers wrote.

A shorter infusion time, the team noted, could help reduce the high burden of treatment for people with Fabry. Among the treatments approved to date for the rare genetic disorder is enzyme replacement therapy, known as ERT, which generates the non-functional or missing enzyme. There are two forms of ERT now available in many countries: Replagal (agalsidase alfa) and Fabrazyme (also available as a biosimilar in Japan).

Fabrazyme, approved in the U.S. in 2003, is administered by infusion into the bloodstream (intravenously) every two weeks, at a dose of 1mg/kg of body weight.

In the U.S., infusions lasting at least 90 minutes are recommended for all patients, and both the U.S. and European Union labels mandate infusions not exceeding 15 mg/hr for patients weighing less than 30 kg (around 66 pounds). However, the safety and tolerability of infusion times that are less than 90 minutes remain to be established.

Read full story: bit.ly/fabrazymetime

Appeals court overturns dismissal of 2020 Fabrazyme lawsuit

Fifteen years after Genzyme began rationing Fabrazyme due to a shortage caused by contamination at a manufacturing site- launching many years of litigation - the U.S. Court of Appeals has ruled a plaintiff group has legal standing to assert its claims. This reverses a prior ruling in Massachusetts.

Read full story: bit.ly/fabrasuit

Nurses, patients work together to make home infusions successful

By Susanna VanVickle
FabryDiseaseNews.com

(Nov. 1, 2023) The two clinical sites where our kids began their ERT for Fabry were staffed by top-notch nurses. My kids enjoyed snacks and entertainment while skilled hands worked IVs into their veins.

Making the decision to do home infusions was a natural choice, however. The kids couldn't wait for the day when they could stay in their pajamas and get their infusions in the comfort of their own home.

My now 21-year-old twins and their sister, Marisa, 13, have together had more than 200 infusions in the past four years as part of their treatment for Fabry disease, and it's fair to say no two infusions have been the same. Even the setting has changed — major hospitals, small student exam rooms on college campuses, outpatient clinics, dorm rooms, and our living room couch.

Once, an infusion was finished up at a restaurant!

Read full story: bit.ly/Home-Infusions

Court strikes down copay accumulators

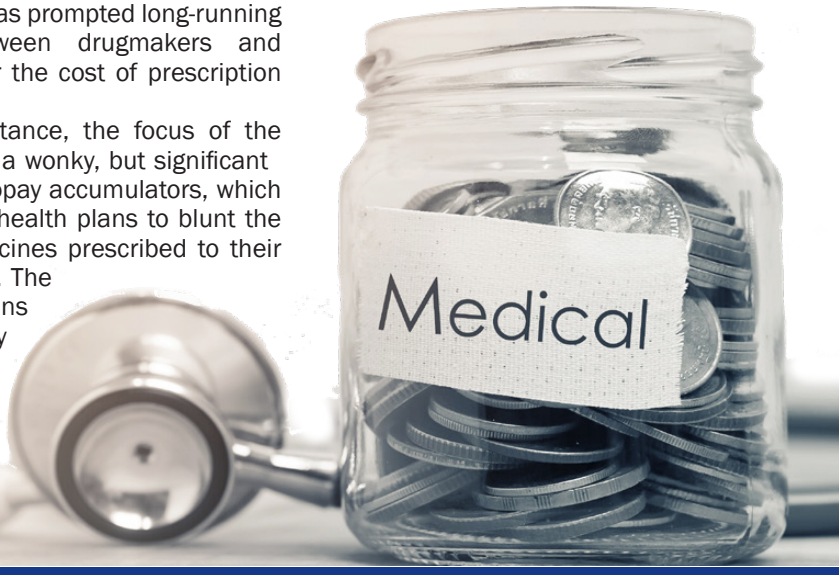
By Ed Silverman
StatNews.com

(Oct 2, 2023) A U.S. judge has struck down a Trump administration rule that allowed health insurers not to count copay assistance offered by drug companies toward out-of-pocket costs, a victory for advocacy groups that argued the rule harmed patient health.

At issue is the complex and often opaque health insurance system in the U.S., which has prompted long-running battles between drugmakers and insurers over the cost of prescription medicines.

In this instance, the focus of the case was on a wonky, but significant tool called copay accumulators, which are used by health plans to blunt the cost of medicines prescribed to their beneficiaries. The story begins with copay assistance programs, which drugmakers have offered for years to help patients afford medicines by mitigating out-of-pocket and deductible costs. Health plans counter that such assistance—generally, in the form of copay coupons or cards—is just a marketing tool used to direct consumers to higher-priced drugs, which eventually raise costs to the entire health care system.

Read full story: bit.ly/copay-accum



FDA clears uniQure trial of AMT-191 gene therapy

US-based Phase 1/2a trial expected to begin in 1st half of 2024

By Lindsey Shapiro, PhD
FabryDiseaseNews.com

(Dec. 1, 2023) The U.S. Food and Drug Administration (FDA) has cleared a Phase 1/2a trial to test uniQure's gene therapy candidate AMT-191 in a small group of people with Fabry disease.

UniQure says it expects that enrollment in the U.S.-based trial—which will recruit six patients in all—will begin in the first half of next year. The company sought approval of the clinical trial via an investigational new drug application (IND).

"AMT-191 has the potential to be a differentiated gene therapy for the one-time treatment of Fabry disease," Walid Abi-Saab, MD, uniQure's chief medical officer, said in a company press release.

"We have designed the Phase I/II study to provide dose-ranging biomarker data as rapidly and cost-effectively as possible, and we look forward to enrolling our first patient in the first half of 2024," Abi-Saab said.

AMT-191 is a one-time gene therapy designed to deliver a working version of the GLA gene directly to liver cells, enabling them to continuously produce their own healthy Gal A enzyme.

It is packaged into a viral carrier called adeno-associated virus 5 (AAV5), which helps the therapy be taken up by liver cells but is modified so as not to cause disease. It is the same AAV5 technology used in Hemgenix, a recently approved gene therapy for hemophilia B originally developed by uniQure.

The company previously had been developing another gene therapy candidate, known as AMT-190, as a potential Fabry disease treatment. AMT-190 had aimed to provide patients with alpha-N-acetylgalactosaminidase, or NAGA, an enzyme similar to Gal A.

In September 2020, however, uniQure selected AMT-191 instead as its lead gene therapy candidate to be advanced into IND-enabling studies. Preclinical studies comparing the gene therapies found that AMT-191 led to more robust and sustained increases in GLA activity and functional improvements, according to the company.

Read full story: bit.ly/uniQure-Trial

4DMT, FDA agree to lift clinical hold on Fabry gene therapy

By Steve Bryson, PhD
FabryDiseaseNews.com

(Nov. 3, 2023) 4D Molecular Therapeutics (4DMT) has reached an agreement with the U.S. Food and Drug Administration (FDA) to lift the hold on a U.S.-based clinical trial testing 4D-310, an investigational gene therapy for Fabry disease.

The hold was based on safety data from the company's INGLAXA Phase 1/2 trials. One is taking place in the U.S. (NCT04519749) and another in Taiwan and Australia (NCT05629559).

Among the six patients dosed in both trials, three developed atypical hemolytic uremic syndrome (aHUS), a rare disorder marked by the destruction of red blood cells that carry oxygen in the bloodstream.

To address this, 4DMT launched a single safety study involving nonhuman primates to evaluate 4D-310 combined with an immunosuppressive regimen using rituximab and sirolimus (R/S) to lower the risk of aHUS and toxicity-related side effects. The company plans to submit the results to the FDA by mid-2024.

The INGLAXA trial protocol has also been modified to minimize aHUS risk, including by implementing the R/S immunosuppressive regimen.

The company is conducting the single study in monkeys to demonstrate that switching the immunosuppressive regimen from prednisone to R/S is safe and has a good distribution in the body.

4DMT is continuing to examine efficacy data from the INGLAXA clinical trial program. Interim data, including results from heart biopsies, after 1-1.5 years of follow-up for all six patients, are expected in the first quarter of 2024.

Read full story: bit.ly/Fabry-4D-310

ST-920 gene therapy lowers Fabry disease severity in trial

By Marisa Wexler, MS
FabryDiseaseNews.com

(Feb. 9, 2024) The investigational gene therapy ST-920 (isaralgagene civaparvovec) has been well tolerated so far among adults with Fabry disease in the Phase 1/2 STAAR clinical trial, according to new interim data from its developer Sangamo Therapeutics.

One-year data from more than a dozen patients indicate ST-920 stabilized kidney function, reduced disease severity, and improved quality of life. The findings were shared as an oral and poster presentation at the 20th Annual WORLDSymposium in San Diego.

"We remain encouraged by the emerging safety and efficacy data supporting the potential durable benefit that ST-920 could offer patients with Fabry disease as a convenient single-dose treatment option," Lisa Rojkjaer, MD, chief medical officer of Sangamo, said in a company press release.

Dosing in STAAR is expected to finish soon and Sangamo is gearing up for a Phase 3 trial that could serve to aid ST-920's approval.

At the time gene therapy was given, 13 of the study participants were on enzyme replacement therapy (ERT). As of the latest follow-up, 12 of them have stopped ERT and none have had to restart it. Alpha-GalA levels have stayed in the supraphysiological range for all but one of the 12, whose enzyme levels are normal.

Four patients had such dramatic changes in MSSI that their overall category of disease classification changed. One patient who'd had severe disease was reclassified as moderate, while three who'd had moderate disease were changed to mild.

Read full story: bit.ly/st920-trial

Fabry gene therapy improves heart health

4D-310 delivers functional GLA copy to heart muscle cells

By Steve Bryson, PhD
FabryDiseaseNews.com

(March 15, 2024) Measures of heart function improved one to two years after treatment with 4D-310, an investigational gene therapy for Fabry disease, according to new trial data from 4D Molecular Therapeutics (4DMT), the therapy's developer.

"We are pleased to see 4D-310 continue to consistently demonstrate clinical activity across multiple important cardiac endpoints, including cardiac function, exercise capacity and quality of life," Robert Kim, MD, chief medical officer of 4DMT, said in a company press release.

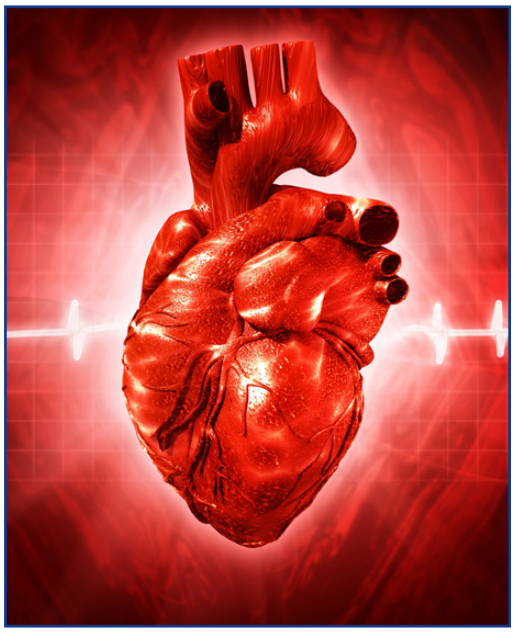
The data were presented in a late-breaking session at WORLDSymposium 2024 in San Diego.

Fabry disease is caused by mutations in the GLA gene and result in a deficiency in the alpha-Gal A enzyme, which breaks down fatty molecules, primarily globotriaosylceramide (Gb3 or Gl-3). Without sufficient alpha-GLA, the molecules build up to toxic levels, causing tissue and organ damage, particularly in the kidneys and heart.

Enzyme replacement therapy (ERT) is the standard Fabry treatment and delivers a functional, lab-made alpha-Gal A enzyme into the bloodstream. Despite being able to improve kidney function, ERT doesn't fully address impaired heart function.

"Current therapies do not adequately address Fabry-related cardiovascular manifestations, and cardiovascular disease is the most common cause of death in these patients," Kim said.

Read full story: bit.ly/Fabry-Heart



GENETIC BRIEFS

Cardiovascular genetics solve medical mystery

A writer for *UConn Today* found himself the subject of a medical mystery, when doctors at UConn Health couldn't figure out the cause of his unusual symptoms.

"What started as a trip to a walk-in clinic turned into a hospitalization and became a medical mystery that went unsolved for nearly four months," wrote alumnus Christopher DeFrancesco in a Feb. 29 article on the college's website.

After a simple check of shoulder pain revealed concerning heart problems (which he didn't even notice), cardiologist Agnes Kim first mentioned Fabry among several possible causes... eventually leading to a diagnosis at age 50.

Read full story: bit.ly/UConnMystery

New FDA committee could boost field

On Dec. 14, the U.S. Food and Drug Administration announced its creation of a new advisory committee related to genetic metabolic diseases. The decision brings forward a field that has long languished, as such conditions are complex, hard to even diagnose and even harder to treat.

The field has been driven forward by companies such as diagnostic firm Centogene, and gene therapy specialists, including Uniqure and 4D Molecular Therapeutics. Maze Therapeutics in the U.S. and Scenic Biotech in the Netherlands are developing drugs for lysosomal storage disease.

Read full story: bit.ly/Committee-GMD

Fabry pain affects each of us uniquely

By Jerry Walter
FabryDiseaseNews.com

(Dec. 20, 2023) My inspiration for writing this article about Fabry disease pain came from the graphic representation of pain shown above.

Neuropathic pain is a common manifestation of Fabry that is studied and written about frequently, but this illustration is unique. It was shared with me by the pharmaceutical company Sanofi in my role as president of the National Fabry Disease Foundation.

This depiction of Fabry pain is a product of a recent patient advisory board meeting that Sanofi held to learn more about patients' experiences with pain. Attendees' thoughts were documented and transformed into this relatable visualization.

Understanding Fabry pain is an important ongoing area of study that began decades ago. It remains a frequent topic of discussion and research because it significantly affects many individuals' daily activities and quality of life. Hundreds of peer-reviewed scientific articles have been published about various aspects of this symptom.

For many people, Fabry pain is just annoying and minimally disruptive. For others, it worsens to become severe and debilitating. Understandably, chronic pain can affect a person's mood, demeanor, and personality, as well as their school, work, and social life..

Through the scientific literature available, talking with others, social media discussions, and my personal experiences, I've learned that manifestations of Fabry pain are complex and often difficult to treat. The type, location, severity, age of onset, triggers, treatments, and effects on quality of life vary widely among individuals.

One article published in the journal *Pain* describes neuropathic Fabry pain as "burning, searing, squeezing, pressing, dragging, prickling, electrifying, tingling, sharp, and sore." Other articles include additional unpleasant descriptions.

The graphical depiction is not all-encompassing, but I think it's a good representation of the diversity and complexity of Fabry pain and its effects.

My pain changes often and I often experience various types simultaneously. Because I get many random, sharp, shooting pains all over my body, I can especially relate to the image of the voodoo doll in the center of the graphic. My version of the concept is that I feel like I have microscopic gremlins — those mischievous creatures of folklore in a super downsized form — running around my peripheral nervous system with their little bags of tools to stab, poke, pinch, cut, and sting me. Their modus operandi is to hit and run as they disrupt my comfort and peace of mind at their whim. My pain experiences primarily fall into three categories:

1. Frequent, random, sharp, shooting pains are delivered by the gremlins. Most of my pain is invisible to others, but these manifestations may be visible in instances of me flinching, grimacing, or clenching my teeth with the occasional quiet yelps.

2. Chronic, widespread flu-like achiness sometimes affects my desire or ability to participate in various activities. I've had some level of general achiness nearly every day for many years. It worsens with strenuous activity, temperature and humidity changes, and fevers. This affects my qualify of life the most.

3. The sensations of burning, tingling, pricking, and numbness in my extremities are known as acroparesthesia. This is often one of the earliest reported symptoms of Fabry. When this occurs, reduced circulation causes my hands to change colors.

Read full story: bit.ly/Fabry-Pain

Complementary health increasing in popularity to manage pain

The use of complementary health approaches among patients in the U.S. increased between 2002 and 2022, parallel to an increase of reported pain prevalence, according to findings published in *JAMA*. There has been a growing body of evidence to support their safety and efficacy for paint, such as acupuncture, guided imagery and/or progressive muscle relaxation, massage, naturopathy, and yoga.

Read full story: bit.ly/CHAs

In the face of pain, expressing gratitude grows our happiness

By Susanna VanVickle
FabryDiseaseNews.com

(Nov. 28, 2023) Whether it's the breath in our lungs, the glow of the sun, the changing of seasons, a smile from a stranger, a call from a friend, music that moves us, or beauty, freedom, family, and faith, the present moment is ripe with goodness. Yet often that goodness is marred by the annoyances or pains that vie for our attention.

In my family's Fabry disease story, for example, bona fide blessings abound. I have the same Fabry mutation as my children, but the disease hasn't manifested in me while it has in three of them. We've found answers and treatment early in my kids' lives, and they've been under the care of some of the world's leading experts on Fabry disease. When we focus on these favors, happiness prevails.

There are moments, however, when sunshine is fleeting, it's hard to focus on blessings, and living with Fabry is exasperating. Can happiness be found when storm clouds roll in?

When my kids' gastrointestinal pain soars, I want to wring my hands because there's nothing I can do to stop it. When my adventurous, enterprising sons pass up an opportunity because of how taxing the heat would be on their bodies, my heart breaks for them. When my daughter laments her experience with enzyme replacement therapy, I want to call it quits.

How can happiness and goodness coexist with suffering and lack? The answer in my life has been to give thanks. We can choose an attitude of gratitude. Happiness is not an action we can take, yet it can grow in us as the result of the action of giving thanks.

As I take inventory of my home and family, my gratitude list expands, noting strong bodies and minds, an understanding of this rare disease, early Fabry diagnoses, amazing area doctors, treatment options, assistance funds, modern medical facilities, and the online community we've found.

Read full story: bit.ly/Grow-Happiness

Identifying small fiber neuropathy may aid in timely Fabry treatment

By Lindsey Shapiro, PhD
FabryDiseaseNews.com

(Jan 12, 2024) Identifying small nerve fiber involvement early in Fabry disease (FD) could be critical to enabling timely diagnosis and treatment, according to a recent report.

An array of clinical, sensory, and nerve function tests were used to identify signs of small nerve fiber neuropathy (SFN), or symptoms of nerve damage, in more than half of the people evaluated for suspected Fabry disease in the study.

Ultimately, SFN was critical for enabling the start of enzyme replacement therapy (ERT) for some patients, including those who were or were not definitively diagnosed with the disease.

Altogether, the findings highlight a "specialized and detailed examination" is needed to identify small nerve fiber involvement in Fabry patients, according to researchers.

In the study, the scientists conducted a retrospective analysis of clinical data from patients evaluated for possible Fabry disease, looking specifically for signs of small nerve fiber dysfunction.

According to Fabry disease diagnostic criteria, 19 people could be definitively diagnosed with the disease, 13 of whom had evidence of SFN.

Among these 19 patients, 16 were treated with ERT, as were another three people who did not have a definitive diagnosis. For all three patients without a diagnosis, and for two with one, identification of SFN was a critical factor for deeming them eligible to receive ERT.

"This is very important because when treatment is initiated even before renal function starts to decline, ERT may improve small nerve fiber function and help decrease pain," the researchers wrote. "The early initiation of ERT as well as the good supportive pain management using adjunctive therapies may improve patient's quality of life.

Read full story: bit.ly/neurop-early

Cannabis sole help for man's nerve pain

By Patricia Inácio, PhD
FabryDiseaseNews.com

(Dec. 8, 2023) A man with Fabry disease experienced an easing of neuropathic pain, or nerve pain, with medical cannabis after failing to respond to standard therapies, a case study reports.

Neuropathic pain is one of the most distressing symptoms for Fabry patients and may not ease with standard treatments such as enzyme replacement therapy (ERT). This type of pain is prevalent in Fabry disease, affecting 81.4% of male patients and 65.3% of female patients, according to researchers.

The patient had begun treatment with Fabrazyme, an ERT, together with olmesartan, a medication for high blood pressure.

After treatment for one year, while some symptoms were eased, the patient still experienced severe burning pain in his extremities, which took a toll on his sleep and quality of life.

Over the next three years, several medications were prescribed by a neurologist, but none eased the patient's pain. Among these were anti-epileptic and anti-convulsant medications.

The man also took paracetamol, an over-the-counter painkiller used in emergency situations for acute pain attacks.

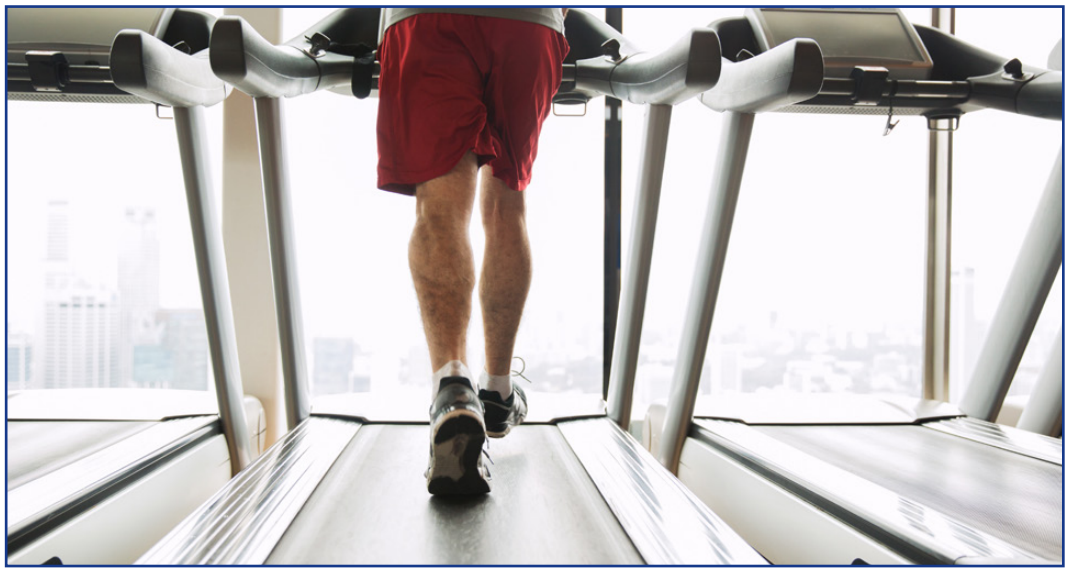
By 32, the patient was referred to a pain management specialist. He was prescribed oral medical cannabis (cannabidiol 12% and tetrahydrocannabinol 8%), two of the most abundant cannabinoids found in the cannabis plant. The dose began at 100 mg per day, and increased to 500 mg per day.

One month after treatment, the patient reported easing of pain, without significant side effects, as assessed by the Brief Pain Inventory questionnaire. This relief, particularly of nocturnal pain, was maintained for one year after treatment.

Read full story: bit.ly/NervePain-Cannabis

13 key ways I stay healthy with Fabry

By Jerry Walter
FabryDiseaseNews.com



(Nov. 22, 2023) This week, I turned 69 years old. As a man with classic Fabry disease, I consider this achievement both significant and unexpected. I'm still getting used to the idea that living this long is possible for people like me, after believing that it wasn't for most of my adult life. Medical literature indicates a shorter life span for Fabry patients, and five of my family members died between the ages of 37 and 51 from complications related to the disease.

I discovered a resource from the American Heart Association called Life's Essential 8, which details ways we can improve and maintain cardiovascular health to help us enjoy a longer, healthier life. Life's Essential 8 outlines eight steps people can take to live a healthier lifestyle:

- **Eat better**
- **Be more active**
- **Quit tobacco**
- **Get healthy sleep**
- **Manage weight**
- **Control cholesterol**
- **Manage blood sugar**
- **Manage blood pressure**

These eight steps are important for everyone, but especially for people with Fabry disease, who are at a greater risk for heart disease, kidney disease, lung disease, and stroke. In addition to minimizing these life-threatening risks, these practices can ease disease manifestations that hurt our quality of life.

For me, the following steps also have been critical to my overall health and well-being:

- Take a Fabry-specific treatment
- Adhere to the schedule of tests recommended for people for Fabry, such as periodic cardiac and cranial MRI
- Stay well hydrated
- Manage albuminuria (elevated albumin in the urine) and proteinuria (elevated protein in the urine)—both of which may indicate kidney issues—with an adjunct therapy, in addition to my Fabry-specific treatment
- Manage lower leg edema, a common issue in Fabry, by wearing compression socks.

Some of my success is just me being fortunate, while other elements require effort. For example, getting enough quality exercise and consistently eating healthily can be difficult to accomplish with my busy work schedule. Many people without Fabry disease probably face the same issues, but a chronic illness can add a layer of complexity.

I hope sharing my essential steps helps others to develop their own and to live better and longer lives.

Read full story: bit.ly/Fabry13

Beloved music store owner succumbs to Fabry

(Feb. 20, 2024) William "Bill" Higgins, a self-taught guitar player who founded an iconic musical instrument shop in Catonsville, Maryland, died Wednesday of Fabry disease. He was 81.

According to his obituary in the Baltimore Sun, Higgins had no formal music training and picked up popular songs by ear. He placed an ad in a local newspaper promoting his services as a music teacher and soon found that he needed a central location.

The business, which was run by Higgins

and his wife, Nancy, opened in 1965 and grew to become a retailer of well-known instrument-makers. He soon won the Gibson, Fender and Peavey accounts. Higgins expanded his customer base by offering credit to young musicians.

"Bill had basic entrepreneurial street smarts," said longtime friend Al Cunniff. "He had no business degrees or college. In the midst of that era of rock music—the time of The Beatles and local bands that played at Catholic Youth Organization dances—Bill was there at the right time."

Survivors include his wife of 57 years, Nancy Lee Baker Higgins; a son, Brian Paul Higgins, and two daughters, Tracey Higgins Kern and Jamie Lee Reese.

Read full story: bit.ly/billsmusic

ACKNOWLEDGEMENTS

THANK YOU to the following supporters, who made gifts from Oct. 20, 2023, through April 18, 2024.

Contributions in Honor of

Jason Frett by Russell Gash
Amy Young by Russell Gash

Contributions in Memory of

Patricia Bellamy by Daniel Schrader
Kenny Bellamy by Daniel Schrader
Janity Blea by Lori Wise
Frank Finn by Lori Wise
Alan & Mary Larese by Lori Wise
Sheryl Plutt by Lori Wise
Mietek Weglewski by Nancy Levy

Other Valued Contributors

Anonymous
Alice Bordelon
Carol Dauria
Michelle Good
Barbara Griffey
Trinh Kelleher
Cheri Peon del Valle
Peter & Christine Radasch
Linda & Pete Todsen
Sherry Tranchemontagne
Barbara Wing

2023 5K Contribution in Honor of

Brandi Johnson by Nancy Rice

2023 5K Contributions in Memory of

Scott Athanasiou by Lisa Bunszel, Carol Dauria, Kristin McMenomey
William Wayne Butler by Christine Butler
Warren Halley by Anonymous
Alan Larese by Lynn Boudreau

2023 5K Other Valued Contributors

Anonymous
Fred Battah
Susan Bowland
Martha Buchanan
Jeff Cairns
Jean Campbell
Kathryn Carlson
Joseph Channells
Carol Dauria
Carol Demeo
Barbara Earley
Shirley Fester
Heather Flater
Lee Goddard
Jennifer Gonzalez
Michael & Elizabeth Gnidovec
Barbara Griffey
Lisa Hall
David Higgins
Jay Johnson
Stephen Kagan
Trinh Kelleher
John King
Tamara Klein
Julie Kolberg
Molly McCarthy
Cheri Peon del Valle
Linda Quaranta
Kristel Renick
Ashley & Randall Rowland
Doris Schappe
Donna Schultz Van Fleet
Lonna Sexton
Linda & Peter Todsen
Sherry Tranchemontagne
Ian Vonesh

2024 Sponsors FSIG Expert Fabry Conference

4D Molecular Therapeutics
Accessia Health
Amicus Therapeutics
Centogene
Chiesi Global Rare Diseases
Data Registry Services
Lysosomal & Rare Disorders Research & Treatment Center
MedPanel
Octant Biosciences
Sanofi
Spark Therapeutics
The Assistance Fund
uniQure

2024 Donations Newborn Screening Testing for Tots

Natalie Baumann
Margaret Clayton
James & Nancy Edwards
Judith English
Thomas & JoAnn Jones
Tim & Kelly Mahon
John & June Rosenbalm
Becca Steifle
Shay Willamon
Cyndee York

2024 Newborn Screening Testing for Tots Corporate Donations

Amicus Therapeutics
Chiesi Global Rare Diseases
Clayton Tile Distributing
GE Foundation
PHX Holding
Sanofi

2024 Donations Fun Run/Walk

James Brown
Ailleen Calantog
Antwaun Cook
Carol Dauria
Carlee Domke
Barbara Earley
Heather Flater
Mary F Hoffman
Trinh Kelleher
Cathy Krasnow
Antonia Lane
Cheri Peon del Valle

Kelly Ranallo
Ashley & Randall Rowland
Eve Rubell
Sherry Tranchemontagne
Biliana Veleva
Lisa Wright

2024 Sponsors Fun Run/Walk

Amicus Therapeutics
Chiesi Global Rare Diseases
Sanofi
uniQure

Matching Funds Contributions

Ameriprise Financial via Benevity Community Impact Fund by Trinh Kelleher
Blue Shield of California via Benevity Community Impact Fund
Costco via frontstream by Michelle Good
Disney via Charities Aid Foundation America via CyberGrants
Fidelity Charitable
Microsoft via Benevity Community Impact Fund by Daniel Schrader
Pfizer via Benevity Community Impact Fund
The GE Foundation Matching Funds

Corporate & Industry Sponsors

4D Molecular Therapeutics
Amicus Therapeutics
Chiesi Global Rare Diseases
Community Shares of Colorado
Data Registry Services
Engage Health

EveryLife Foundation for Rare Diseases
Facebook via various fund-raisers
GE Foundation
Lysosomal & Rare Disorders Research & Treatment Center
M6P Therapeutics
MedPanel
Microsoft
Mightycause Charitable Foundation
Network for Good
Oak Foundation PayPal Giving Fund
PayPal/Venmo
Pillar Patient Advocates Rare Patient Voice
Sanofi
Spark Therapeutics
The Assistance Fund
UK Online Giving Foundation
uniQure
United Way
Vertex Pharmaceuticals Rewards
Your Cause

MAKE YOUR GIFT TODAY!

108 NE 2nd Street
Suite C
P.O. Box 510
Concordia, MO 64020

www.fabry.org

The 11th Annual FSIG Expert Fabry Conference was held Feb. 9-11 in sunny San Diego, CA. Speakers included Dr. Hopkin, Dr. Wallace, Dr. Jefferies and Dawn Laney, among many other Fabry advocates. The weekend was packed with disease education, but what seemed to be the favorite was recognizing four amazing caregivers and one medical professional. This year's Best Caregiver award went to Tabitha Dowdy, and Best Doctor award to Dr. Hammad Bokhari.

FSIG Connection
The Official Newsletter of the Fabry Support & Information Group
108 NE 2nd St.
PO Box 510
Concordia, MO 64020-0510

Return Service Requested

Non-Profit
US Postage
PAID
Permit No 510
Concordia MO

FSIG Connection

ONE-KIDNEY *Warriors*

ALSO IN THIS ISSUE